C19orf12 gene

chromosome 19 open reading frame 12

Normal Function

The *C19orf12* gene provides instructions for making a protein whose function is unknown. The protein is found in the membrane of cellular structures called mitochondria, which are the cell's energy-producing centers. Researchers suggest that the C19orf12 protein plays a role in the maintenance of fat (lipid) molecules, a process known as lipid homeostasis.

Health Conditions Related to Genetic Changes

mitochondrial membrane protein-associated neurodegeneration

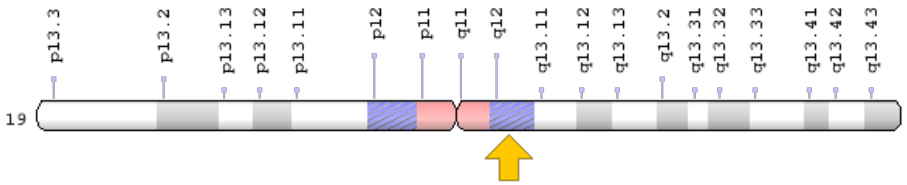
At least 28 mutations in the *C19orf12* gene have been found to cause a condition known as mitochondrial membrane protein-associated neurodegeneration (MPAN), which is characterized by movement and neurological problems that gradually worsen. Affected individuals also have an abnormal accumulation of iron in certain regions of the brain. The gene mutations that cause this condition change single protein building blocks (amino acids) in the C19orf12 protein or lead to an abnormally short protein. These changes likely reduce or eliminate the function of the protein. One *C19orf12* gene mutation, which deletes 11 DNA building blocks (nucleotides) from the gene, is found in people of Polish descent who have the condition. This genetic change leads to production of an abnormally short protein, which is quickly broken down. It is unclear how loss of C19orf12 protein function leads to the signs and symptoms of MPAN. Researchers are working to determine whether there is a link between problems with lipid homeostasis and brain iron accumulation or how these abnormalities might contribute to the features of this disorder.

C19orf12 gene mutations can cause a spectrum of related conditions with some but not all of the characteristic features of MPAN. For example, some affected individuals have movement problems such as muscle stiffness (spasticity) but not iron accumulation in the brain; these individuals are considered to have a condition called hereditary spastic paraplegia type 43.

Chromosomal Location

Cytogenetic Location: 19q12, which is the long (q) arm of chromosome 19 at position 12

Molecular Location: base pairs 29,698,886 to 29,715,789 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- NBIA3
- NBIA4
- protein C19orf12
- SPG43

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28C19orf12%5BTIAB%5D%29+OR+%28%28NBIA4%5BTIAB%5D%29+OR+%28SPG43%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- CHROMOSOME 19 OPEN READING FRAME 12
  http://omim.org/entry/614297

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=C19orf12%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=25443
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/83636
- UniProt
  http://www.uniprot.org/uniprot/Q9NSK7

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